Public engagement with genomic medicine: a summary of town hall discussions

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Abstract

Engaging with the public for their input about genomic medicine is critical before it is implemented into routine healthcare practice. In order to inform discussion and planning for the introduction of genome sequencing into clinical care in an Eastern Canadian province, we implemented a program of public engagement activities. Here, we report a qualitative summary of two town hall discussions utilizing a hybrid information–consultation approach with 20 residents of the province of Newfoundland and Labrador, Canada. Discussion revealed largely positive attitudes towards genomic medicine; however, critical reflection around informed consent models, the return of sequencing findings, and access to qualified healthcare professionals revealed numerous public concerns. Public support will be important to realize the potential benefits of genomics and precision medicine to health outcomes. Our findings highlight public concerns that must be addressed in educational and informed consent documents related to sequencing. Town hall attendees endorsed ongoing public education and awareness-building initiatives which could help foster transparency and trust as genomics is integrated into healthcare systems.

Keywords Public engagement · Genomics · Sequencing · Personalized medicine · Consultation · Qualitative

Introduction

Personalized medicine approaches have the potential for improving health. Examples include in the diagnosis and treatment of rare diseases (Yang et al. 2013; Jacob et al. 2013; Green and Biesecker 2014) and pharmacogenomic testing to guide therapeutic decisions in oncology and other clinical

areas (Pulley et al. 2012; Manolio et al. 2013; Gottesman et al. 2013; Rosenman et al. 2017). Practice guidelines for genomic applications continue to emerge. For example, pharmacogenomic drug therapy guidelines are now available (Gottesman et al. 2013; Rosenman et al. 2017; Crosslin et al. 2015). The American College of Medical Genetics and Genomics (ACMG) recommended a list of 56 actionable genes (revised to 59) to be analysed for disease risk variants, including those unrelated to the initial indication for testing (Kalia et al. 2017; Green et al. 2013). As the cost of sequencing decreases, whole exome or genome approaches will likely become more common in routine practice (Mackley et al. 2017).

All of this implies that increasing numbers of patients will be presented with sequencing decisions in clinical care. The numbers are significant: globally, estimates suggest that almost two billion people will have had their genomes sequenced by 2025, highlighting the need to involve the public in discussions and critical reflection about genomics (Nunn et al. 2019).

Despite the potential to positively impact health, next-generation sequencing (NGS) technologies, such as whole exome or whole genome sequencing (WES and WGS, respectively), raise ethical and scientific issues. The amount and

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complexity of sequencing data generated, the actionability of results, and the management of incidental or secondary findings (Mackley et al. 2017; Korol et al. 2013; Schwartz et al. 2018) are just some of the concerns. As NGS moves from research and rare disease contexts into routine healthcare, stakeholder engagement will be critical for successful implementation (Hartzler et al. 2013). In keeping with calls to meaningfully engage patients and the public in health research and genomics initiatives (Hartzler et al. 2013; Etchegary et al. 2017; Abhyankar and Etchegary 2019; Nunn et al. 2019), this paper presents a summary of two town halls designed to gather public input that might inform the development of genomic medicine in a provincial healthcare system in Canada.

The context for public engagement

Proactively exploring public expectations for, understanding of, and concerns about genomic medicine is informative to guide its implementation into routine practice. In the Eastern province of Newfoundland and Labrador (NL), Canada, healthcare services are provided through four regional health authorities. These include Eastern Health (covering half the population and the site of the Provincial Medical Genetics Program (PMGP)), and the more rural Central, Western, and Labrador-Grenfell Health authorities. Sequencing tests are ordered by medical geneticists in the PMGP, with a minority by oncologists. Samples are sent outside the province for testing; however, the procurement of a next-generation sequencer and ongoing policy and planning discussions over the last 2 years suggest sequencing will soon be carried out locally. Thus, increasing numbers of patients will be making genome sequencing decisions in the jurisdiction before too long.

As part of planning for the introduction of genome sequencing into our healthcare system, we initiated a series of public engagement activities in late 2018 that included (1) convening a public advisory council on genomics; (2) public town hall sessions; and (3) an online provincial survey. In all public engagement activities, we explored how members of the public think about genome sequencing in order to plan how to better support them in making sequencing decisions and inform the development of local testing policies and informational resources.

We previously reported on the creation of the public council (Abhyankar and Etchegary 2019). Briefly, a 12-member council was convened for 1 year to discuss the introduction of sequencing into care. The council had largely positive attitudes about sequencing and suggested it could have substantial benefits, including reducing healthcare costs through more appropriate drug therapy and enabling diagnoses more efficiently. But the council also identified areas of concerns, including patients' and healthcare providers' ability to truly understand sequencing and the range of possible results, as well as the privacy and protection of genomic data (e.g., the

physical security of data storage facilities and data access protocols). The council suggested ongoing opportunities for public awareness building and discussion, including public consultation sessions and a larger, more representative provincial survey.

While survey findings are reported separately (Etchegary et al. 2020), they are summarized in brief here. An online provincial survey of over 900 residents revealed high levels of interest in WGS, with almost 60% definitely interested and over 20% somewhat interested. A majority (~86%) also indicated interest in pharmacogenomic testing specifically. Issues respondents identified as very important to sequencing decisions included knowing the familial implications of testing, whether treatment was available for conditions tested and knowing who could access genomic information (e.g., data access and privacy concerns). Most respondents (73%) also agreed they would value support from a healthcare provider when interpreting sequencing results.

In this paper, we report the results of the first two public consultation sessions.

Materials and Methods

Public consultation approach

A variety of methods, from surveys to deliberative citizen juries, have been used to engage public stakeholders with genomics research (e.g., Lemke et al. 2010). We designed a hybrid information–consultation town hall for our initial genomics consultation sessions, having successfully employed this approach in prior community engagement work, both related to genetics research (Etchegary et al. 2013) and health research more broadly (Etchegary et al. 2017).

Guided by the public advisory council's recommendation (Abhyankar and Etchegary 2019) and results of a provincial survey (Etchegary et al. 2020), we anticipated that many members of the general public would not be overly familiar with genomic terms, such as exome vs. whole genome sequencing, incidental findings, or pharmacogenomics. The first goal, therefore, was to provide information and some common definitions for participants. We also wished to provide time for group discussion and questions on a variety of issues related to the introduction of genomic sequencing in our jurisdiction.

We organized two town halls in the capital city of St. John's, NL, during Spring 2019. St. John's is the site of the Provincial Medical Genetics Program (PMGP) and the site where sequencing will first be offered in practice. Sessions were open to any member of the general public over the age of 18 and were held over the lunch hour at the Faculty of Medicine, Memorial University. As this summary of findings from the engagement activity reflects the use of secondary

data that does not identify attendees, ethics approval was not required for town hall sessions. However, attendees were advised that their participation was voluntary, they could contribute to the discussion as much or as little as they wished, and that no names would be associated with any of the comments raised in town halls in published reports from the work.

Public recruitment

Sessions were advertised over Jan–Feb 2019. In all advertisements, individuals were invited to call a toll-free number to register for the session. There was no compensation provided to those who attended, although lunch was provided.

Announcements and posters advertising the sessions were sent to various community organizations and associations to be advertised through their respective newsletters and websites. For example, the capital city's municipal public website, local hobby groups, a patient advisory council situated within the Faculty of Medicine at Memorial University and a University-wide newswire email distribution list. Members of the research team, as well as the public advisory council, extended personal invitations to their networks as well (e.g., patient partners on other research projects).

Consultation materials

Town halls were designed as a hybrid information–consultation session. First, participants were provided with information about the current use of genomic sequencing in clinical care. This included information on how genomic tests are ordered and by whom, for which typical indications, and the fact that sequence data is not currently returned to the province for storage; rather, only sequencing results are returned in a lab report. We also explained that the acquisition of a sequencer in late 2018 now meant sequencing could be accomplished locally, although many governance and logistic issues were yet to be determined. We expressed our desire to engage with the public early, in a proactive way, to help inform the development of provincial sequencing governance frameworks, privacy, and data policies. We also shared information during the town halls about the public advisory council and its suggestion to provide discussion opportunities for other members of the public. Finally, we shared some of the results of the online provincial survey (Etchegary et al. 2020) to help set the stage for discussion (this PowerPoint presentation is available upon request by email to the corresponding author).

We administered an anonymous post-session survey to gather participants' feedback about the session, any final comments on genomics, as well as broad categories of demographic information (age range, sex, and employment status).

Session facilitation

Town halls were facilitated by HE, with detailed field notes taken by the rest of the team. Sessions began with a welcome to participants, followed by a PowerPoint presentation (15–20 min) that explained the upcoming rollout of sequencing into clinical care and then reviewed some basic genetic concepts (e.g., chromosomes, genes, DNA, exomes, genomes, sequencing). We also used town halls as a knowledge translation opportunity to share some of our sequencing survey findings. Town halls lasted 1.5 hours each.

Following the presentation, discussion was prompted with four questions on a PowerPoint slide:

- (1) What kind of consent models could be used in care and/or research about genomic sequencing?
- (2) What kinds of results should be returned to patients and what do we do with any incidental findings?
- (3) What do you think of genomic data sharing—e.g., allowing other researchers to access your genomic data for large research studies?
- (4) What questions do you have—what would need to be answered for you before you agreed to have your genome sequenced?

Participants were asked to use these as a springboard for discussion, and also to pose their own questions and to comment on anything they wished. It is worth noting that discussion was lively and only the first two of these questions were addressed in the time available. Following the discussion, participants were asked to complete their post-session surveys. The discussion was not audio-taped, but flip charts were used to record key discussion points and specific comments; extensive, sometimes verbatim, notes and participant quotes were recorded by team members (MW, AP, and CS).

Analysis of town hall discussions

Our goal was practical and in line with a qualitative descriptive approach (Sandelowski 2000, 2010; Chafe 2017). We aimed to identify public opinions and concerns about the introduction of genomic sequencing into practice to inform the development of sequencing policies and frameworks. Qualitative description provides a comprehensive summary of an issue and can also generate information about how to improve healthcare practice (Bradshaw et al. 2017). Ultimately, the goal of all our public engagement work is to provide information to guide the rollout of sequencing into our provincial healthcare system in ways that align with the very public that will use the service.

Town hall discussions were coded by two team members (HE and CS) and analysed incorporating principles of constant comparison and qualitative description (Sandelowski

2010; Glaser and Straus 2017). First, comments from participants (as recorded on flip charts, field notes, and the post-session short surveys) were read and re-read independently by the two team members to identify and index emerging categories and themes (Glaser and Straus 2017). Using the principle of constant comparison, all data were first compared within consultation session, before being compared across sessions. For example, notes, flip charts, and surveys were analysed for town hall 1, before moving on to town hall 2. Analysis for each individual session was completed before comparing the data across sessions. Once independent coding was complete, investigators' thematic analysis of the comments was compared and found to be very similar (> 90% agreement); differences tended to be minor (e.g., "lack of resources" vs. "concerns about resources").

Results

Participants

In total, 20 members of the public attended town halls, nine in the first session and 11 in the second. Participants were mostly female ($n = 18$), retired ($n = 13$), with children ($n = 17$). Most ($n = 15$) were 50 years of age or older, with the other five between the ages of 20–40. Six participants were currently employed, while one was a fulltime university student.

Key themes arising from town hall discussions

In both town halls, participants expressed largely positive attitudes towards the idea of genomic sequencing, noting its potential health benefits. Open comments from the post-session survey noted "Ideally, this sounds like an amazing initiative to introduce" and "Sequencing could have big implications for the health of Newfoundlanders and Labradorians." Other potential benefits raised during town halls were earlier diagnosis of disease and ensuring the correct type and dosage of drug therapy for patients.

However, here we wish to highlight themes that arose from the majority of town hall discussion that ensued following the endorsement of sequencing's potential benefits. These themes revealed areas participants noted would need to be better understood before sequencing should be introduced to patients in our healthcare system. These revolved around (1) consent for sequencing, including the return of sequencing findings; (2) access to genomic data—by players both within and outside of the healthcare system; and finally, (3) the need for awareness raising and education about genomics. For ease of presentation, we present these three themes in turn. However, it is clear from town hall discussion and the themes that follow that these ideas are interrelated.

Consent for sequencing

Attendees at both town halls quickly agreed that consent for genomic sequencing was a critical issue and careful thought was needed before sequencing was offered to patients broadly. One participant described genomics as "a huge accelerating train" that was coming whether we are ready or not. Discussion about consent included numerous aspects, as the following section reveals.

Participants quickly pointed out that while many of the elements of obtaining consent for sequencing were similar to any other kind of medical consent (e.g., it should be voluntary and informed), they suggested that consent for sequencing was different on at least three fronts: (1) The current level of understanding of the general public about sequencing, including its varied potential implications; (2) The resources required to obtain consent for sequencing; and (3) What kinds of results to return to participants and how to obtain consent for these.

Understanding and implications

There was wide agreement that most "average" people would have limited experience or understanding of what sequencing is and what its potential implications could be:

It's different to go in and get bloodwork done to test for high cholesterol vs. genomic sequencing. Most people won't understand the implications of getting your genome sequenced, so there should be a consent form to explain all this. –Town Hall 2

The topic of genomics is a foreign concept for a lot of people, so the consent form is going to need to serve a dual purpose – regular consent details and education for patients. –Town Hall 1

The public needs to be made aware of what sequencing/testing is available and to be aware of the consequences of what could possibly be discovered. –Town Hall 1, Post Survey Comment

Exchanges during both town hall discussions revealed numerous questions about the implications of sequencing:

What are the implications psychologically – this is what we don't know. This is why there should be counseling, it's not just a yes or no issue. –Participant, town hall 2
There needs to be an awareness and understanding of the implications – how will you cope with the information? How will you live differently? –Participant, town hall 2

What about the impact on other family members who may not have provided consent or may not want to know about the findings? –Participant, town hall 1

Resources required

Resources implications were commonly raised across both sessions:

And we live in a province with high illiteracy rates. How do you fully explain all the information in a two-minute doctor visit? Can the doctor even explain what all this means? Doctors don't have time.

And do the doctors even understand the implications? –Town hall 2

And what about the resources required to obtain consent? Right now if things are done in a research protocol, there is a strict protocol and time to explain. But the time required for this in healthcare would have huge resource implications. –Town hall 2

The healthcare team should be tasked with saying upfront – we don't know what this means now, but when they do, go back and inform the patient. This is a reasonable way to do things, but do we have the resources to do that? –Town hall 1

A comment from a post-session survey also raised resource issues:

Cost—can we afford to do this? Genetic testing is very limited already. How will it affect healthcare resources—e.g., patients demanding investigative tests?

The return of sequencing results

Discussion centred not only on the kinds of results that could be returned, but the importance of patient control and choice, being advised on the possible types of results *before* sequencing and having access to a knowledgeable person to help interpret results following testing.

The most-often discussed aspect of results was patient control and choice, or as one participant put it quite simply, “People want choice.”

Participants regularly suggested that whatever kinds of results were possible from sequencing must be fully explained to patients upfront during a consent process and patients should have the option of selecting which results they would like returned. Indeed, participants noted the return of incidental findings was one different component of informed consent that might not normally be a part of consent processes with which patients are familiar (e.g., bloodwork or surgery consents).

All participants agreed that results for treatable conditions would likely be preferred as these would be considered “good,” whereas most would probably not want to know they were at increased risk for dementias as there is currently no treatment and these would likely be considered “bad.” However, discussion quickly turned to the difficulty of deciding what is good and bad information to return. This was noted as an ethical issue and patient choices should be respected. In the second town hall discussion, one exchange revealed a group suggestion for framing the return of findings on a consent form (from the flip chart):

“People need to be told upfront – here are the possible results, tell us what information you need or results you want. It could be framed as:

Results could come back here that may cause you worry; some of these disorders could affect your children; some are not treatable, etc. These are the types of results possible (insert check boxes):

- Information about conditions that are treatable (for you and your children)
- Information about disorders that are not treatable
- Information that we really don't know what it means yet

People could then select the types of results they are interested in. Maybe include a question or statement for self-reflection – e.g., the information you receive may affect your quality of life because what we think right now in this moment may not be the case 10 years down the road”.

Attendees noted that some patients could react with fear, worry, or anxiety to sequencing results and it would be important for them to have time to reflect on what results they want back.

Partly for this reason, many participants highlighted the value of having a knowledgeable person to help interpret results. Participants wondered about how the results would be reported back, and in what kind of format. They wondered if a lay person would be able to interpret a results report. One participant noted:

You need someone to interpret it and explain it to the patient. For example, finding out you have Huntington's or Alzheimer's – a patient might die of fright from finding out the potential to have the condition vs. the actual condition. It's easy for the public to misinterpret information – even when we think we want it all back. –Participant, town hall 1

There was no consensus on who should return results. While most acknowledged it would be “nice” if their family doctors could discuss results with patients, they also suggested family doctors wouldn't have time. For participants in both

groups, it was most important that the healthcare professional was “knowledgeable” about sequencing and could correctly interpret results. Nurse practitioners and pharmacists were specifically noted as potential professionals.

Access to sequencing results

Participants were most often likely to grant their family doctors access to their sequencing results over any other healthcare provider. There was an acknowledgment that patients tend to trust their family doctors and perceive them as their primary healthcare provider.

However, discussion was generated from the presentation of our survey findings which showed survey respondents were much less likely to allow pharmacists to access sequencing results. Participants in town halls noted that when it came to certain types of sequencing results (e.g., pharmacogenomics results), pharmacists might actually be a preferred professional given their knowledge of drugs and dosages.

I only recently discovered how much a pharmacist knows over a doctor when it comes to drugs and medication. –Participant town hall 2

While participants generally agreed, some noted they would want the totality of their sequencing results shared with their family doctor, but only relevant, drug-related, sequencing results to be accessed by pharmacists. “Pharmacists already have access to our medications, so it wouldn’t be too much of a leap to access pharmacogenomic results.”

However, it was noted that participants wouldn’t want other people working in pharmacies beyond their pharmacist (e.g., reception staff) to have access to all their genome data.

Outside of the healthcare context, questions were raised in relation to the issue of access to results and the implications this could have for patients and families:

When you think about genetic testing, you think about how this will affect my children or grandchildren. What are the ripple effects on employment opportunities, insurance, travel, etc. –Participant, town hall 2

There may be cases where people don’t want to know that a sibling of theirs is not really a sibling. Who outside the medical community is going to have access to this information? Who can request this information, maybe through legal channels? –Participant, town hall 1

What about insurance companies? If they have access to your sequencing results, can they deny coverage or charge higher premiums. –Participant, town hall 1

This discussion raised the question of whether there was any way to keep sequencing information within the medical community. One attendee noted that once your genome is sequenced, any source would be able to find a reason to access that information. An example was given of a criminal found in the USA based on information of a relative who had used a commercial testing company.

The need for awareness raising and genomics education

Across town halls, attendees frequently raised concerns regarding whether both patients and providers had the knowledge and ability to understand sequencing and interpret results. They suggested awareness raising and education needed to be targeted towards both patients and providers.

Participants suggested that a general public awareness campaign could assist in building awareness about sequencing, including its risks and benefits, before a healthcare provider ever offered it to patients. This would assist both patients and providers gain knowledge about sequencing. It could also help reduce the time needed to obtain informed consent:

This needs to happen before people enter a clinician’s office so they are not ambushed with too much information during an appointment – good, general public education. –Participant, town hall 1

A simple video to inform on the basics of this practice [sequencing], showcase its implications, etc. –Post session survey

Interestingly, participants in the second town hall recalled media coverage of the H1N1 flu outbreak and suggested media attention to sequencing (whether radio, TV, print, social media, etc.) could also help build awareness of new genomic initiatives. They recognized this would require resources and suggested provincial or federal governments should lead this initiative if sequencing is to be introduced into routine clinical care.

Discussion

Amid suggestions that sequencing is moving from strictly research or rare disease contexts into clinical care (Manolio et al. 2013) and in line with growing calls for public engagement (Nunn et al. 2019; Hartzler et al. 2013; Etchegary et al. 2017), we held citizen town halls to explore public knowledge and concerns about genomic sequencing.

While town hall participants were supportive of the potential benefits of sequencing, in line with other research (Etchegary et al. 2020; Hull and Vassey 2018), they were also

able to engage with complex policy and ethical issues raised by sequencing. For example, participants noted concerns with the consent process, including ensuring patients truly understood the implications of sequencing, had access to a knowledgeable healthcare professional to help interpret sequencing results, and that proper protections were in place to safeguard sequencing data access. Results of our provincial online survey were similar, with majorities of respondents ($n = 901$) agreeing that information about data sharing and access, as well as healthcare provider assistance with results interpretation, would be critically important in making sequencing decisions (Etchegary et al. 2020).

Our prior research (Etchegary et al. 2020) and that of others (Hyland et al. 2018; Roberts et al. 2018) revealed knowledge gaps and somewhat unrealistic expectations about genome sequencing. Participants in town halls spoke at length about how important it was that patients understand fully the implications of undergoing sequencing, including the range of possible results. Prior work (Esteban et al. 2019; Wynn 2016) has demonstrated that managing patients' hopes and expectations for genomic sequencing is critically important, not only to promote informed sequencing decisions but also to help avoid inappropriate psychological or behavioural responses to results (e.g., feeling falsely reassured or unduly worried). Our findings and others underscore the need to build public awareness and knowledge about genomics in an effort to promote realistic expectations of sequencing. Town hall attendees suggested this be done before the introduction of sequencing into routine clinical care. Notably, the public advisory council on genomics also made this recommendation (Abhyankar and Etchegary 2019). In this jurisdiction, there is an opportunity for such an information campaign, but it would require the commitment of dedicated funding by genomic service planners. Research funding is also needed to test interventions and the development of patient (and provider) materials to support the use of information related to genomic testing. We believe such investments now would only promote the informed and efficient use of genomic medicine services, perhaps saving dollars in the long term.

Town hall participants' opinions on the return of sequencing findings, including incidental findings, correspond well with the literature. In line with "binning models" (Berg et al. 2011; Clift et al. 2015) that classify incidental findings according to clinical utility, severity, or current understanding, our participants also suggested options be provided to patients upfront according to whether the disease was treatable or not, had implications for other family members, or included a result for which there was limited understanding currently. Participants made a very specific suggestion that no one should decide what kind of results a patient should be returned but the patient; choice was paramount. However, they recommended including a statement that encouraged self-reflection on the kinds of results that were possible, including those for

which there is limited understanding about what the results might mean in the future. This seems a fairly easy suggestion to incorporate on consent forms or education materials and may well encourage deeper thought about sequencing implications; furthermore, it is in line with proposed patient-centred approaches to returning sequencing results (Clift et al. 2015). This suggestion is also important given the substantial possibility of receiving an incidental finding. For example, over 90% of participants undergoing genome sequencing were reported to also have a pharmacogenomic result, while over 80% had a carrier result (Vassey et al. 2017). This suggests that in order to truly make an informed decision about sequencing, patients do need to be fully briefed on the range of possible results.

Respondents in our provincial survey also agreed with the need to fully understand sequencing results and indicated help from a healthcare provider with results interpretation would be expected (Etchegary et al. 2020). Town hall participants, however, noted the "huge" resource implications this would have in practice. There is a well-documented lack of speciality genetic counseling services (Haga et al. 2013; Shickh et al. 2018), and this can be expected to worsen as genomics is integrated into routine clinical care. The systematic testing of alternative forms of pre-sequencing counseling, such as through the use of decision aids (e.g., Shickh et al. 2018), is urgently needed given the lack of capacity in the healthcare system. Town hall participants also recommended that other healthcare professionals, such as nurse practitioners and pharmacists, be included in considering who could talk with patients about sequencing. Study participants suggested that given pharmacists' specialized knowledge in drug therapy, involving them in the provision of pharmacogenomics testing, for example, was a reasonable solution that could help minimize health system usage, particularly within primary care.

Finally, and in line with other research (Genetic Alliance UK 2015) and our survey data (Etchegary et al. 2020), study participants expressed concern over the privacy of their genomic data and access to it from non-medical entities (e.g., insurance companies and law enforcement). They wondered if access could be restricted to only those within their circle of care in the medical community. These findings suggest that any awareness or educational campaigns or informed consent documents about sequencing must be very explicit about what safeguards are in place to protect genomic data, a recommendation also made by our public advisory council on genomics (Abhyankar and Etchegary 2019). This information can be tailored to specific uses of genomic data (e.g., for research or clinical care) and can take into account the local context (e.g., provincial or national privacy legislation) and be informed by local public engagement efforts such as ours when available.

Study findings are limited by the small number of participants represented, the lack of diversity in opinion, and the fact

that sessions were held in only one study location and over the lunch hour. While prior work of the team has found that offering lunch is appreciated by participants in research sessions, the timing of the sessions likely excluded working members of the public who might have preferred an evening session. Members of the public living outside the capital city were also necessarily excluded. Future town halls will be run with additional timing of sessions, and plans are underway to hold virtual events that would allow a greater diversity of the public to attend. Thus, current study findings may not generalize to members of the public from rural areas of the province. Town hall participants were largely retired and female. We cannot know how opinions would differ with younger, working, or male participants. It is likely that participants were those who already had some knowledge about and interest in genomics. However, it is encouraging that findings are in line with our larger online survey of residents that included a more representative sample of residents from the province (Etchegary et al. 2020). Our public engagement approach is just one of many, and a mix of engagement approaches would encourage more diversity in opinion.

These study findings align well with our other engagement efforts, including an online provincial survey and consultation with the public advisory council on genomics. Taken together, these engagement activities help to identify issues important to the public that might be considered in the planning and provision of genomic services and research, including in the design of patient-facing educational and consent materials. Public support will be important to realize the potential benefits of genomics to health outcomes, and our findings highlight areas of concerns for patients that must be addressed in policies and practices related to sequencing. Town hall participants rated the session highly and endorsed ongoing public education initiatives. To that end, we are planning additional virtual public consultation sessions to encourage greater public participation from across the province. We are also engaging in knowledge translation activities to share the results of our public engagement efforts with genomic decision makers in our jurisdiction (e.g., a summary of survey findings has been shared, along with recommendations from the public council). Public input is important to inform decisions about genomics in our jurisdiction, and public engagement is important to foster transparency and trust as genomics is integrated into healthcare systems.

Code availability Not applicable.

Authors' contributions All authors contributed to the engagement session conception and design. All authors contributed to public recruitment for town halls. Material preparation, data collection, and analysis were performed by Holly Etchegary and Charlene Simmonds. The first draft of the manuscript was written by Holly Etchegary and all authors commented on previous versions of the manuscript. All authors read and approved the final manuscript.

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Data availability The PowerPoint presentation used in town hall sessions is available by email from the corresponding author.

Compliance with ethical standards

This article does not contain any research with human or animal subjects performed by the any of the authors. Ethics approval was not required for this public engagement exercise as data collected during town hall sessions are “provided without any identifier or group of identifiers which would allow attribution of private information to an individual” Memorial University, Health Research Ethics Board <https://www.mun.ca/policy/browse/policies/view.php?policy=214>. However, this engagement work followed the principles of the Declaration of Helsinki.

Conflict of interest The authors declare that they have no conflict of interest.

Consent to participate Not applicable.

Consent for publication Not applicable.

References

- Abhyankar S, Etchegary H (2019) Rolling out genomic screening: the Newfoundland and Labrador Public Advisory Council on Genomics (PACG). *BMJ Partnership in Practice Series*. <https://blogs.bmj.com/bmj/2019/12/17/rolling-out-genomic-screening/>
- Berg J, Khoury M, Evans J (2011) Deploying whole genome sequencing in clinical practice and public health: meeting the challenge one bin at a time. *Genet Med* 6:499–504
- Bradshaw C, Atkinson S, Doody O (2017) Employing a qualitative descriptive approach in healthcare research. *Global Qual Nurs Res* 4: 1–8
- Chafe R (2017) The value of qualitative description in health services and policy research. *Healthcare Policy* 12(3):12–18
- Clift K, Halverson C, Fiksdal A, Kumbamu A, Sharp R, McCormick J (2015) Patients' views on incidental findings from clinical exome sequencing. *Applied Translational Genomics* 4:38–43
- Crosslin D, Robertson P, Carrell D, Gordon A, Hannah D, Burt A, Fullerton S et al (2015) Prospective participant selection and ranking to maximize actionable pharmacogenetics variants and discovery in the eMERGE network. *Genome Med* 7:1–10
- Esteban I, Lopez-Fernandez A, Balmana J (2019) A narrative overview of the patients' outcomes after multigene cancer panel testing, and a thorough evaluation of its implications for genetic counseling. *Eur J Hum Genet* 62:342–349
- Etchegary H, Green J, Parfrey P, Street C, Pullman D (2013) Community engagement with genetics: public perceptions and expectations about genetics research. *Health Expect* 18:1413–1425
- Etchegary H, Bishop L, Street C, Aubrey-Bassler K, Humphries D, Vat E, Barrett B (2017) Engaging patients in health research: identifying research priorities through community town halls. *BMC Health Ser Res* 17(1):192
- Etchegary H, Wilson B, Rahman P, Simmonds C, Pullman D (2020) Public interest in whole genome sequencing and information needs: An online survey study. *In press Personal Med*.
- Genetic Alliance UK (2015) Genome sequencing: what do patients think? Patient Charter. <https://www.geneticalliance.org.uk/media/1924/>

- [patient-charter-genome-sequencing-what-do-patients-think.pdf](#). Accessed 16 May 2020
- Glaser B, Straus A (2017) The discovery of grounded theory: strategies for qualitative research. Routledge, New York, NY
- Gottesman O, Kuivaniemi H, Tromp G, Faucett W, Li R, Manolio T, Sanderson S et al (2013) The electronic medical records and genomics (eMERGE) network: past, present, and future. *Genet Med* 15: 761–771
- Green R, Biesecker L (2014) Diagnostic clinical genome and exome sequencing. *N Eng J Med* 370:2418–2425
- Green R, Berg J, Grody W, Kalia S, Korf B, Martin C, McGuire A, Nussbaum RL, O'Daniel JM, Ormond KE, Rehm HL, Watson MS, Williams MS, Biesecker LG, American College of Medical Genetics and Genomics (2013) ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. *Genet Med* 15:565–574
- Haga S, Burke W, Agans R (2013) Primary care physicians' access to genetic specialists: an impediment to the routine use of genomic medicine? *Genet Med* 15(7):513–514
- Hartzler A, McCarty C, Rasmussen L, Williams M, Brilliant M, Bowton E, Clayton E et al (2013) Stakeholder engagement: a key component of integrating genomic information into electronic health records. *Genet Med* 15:792–801
- Hull L, Vassey J (2018) Toward greater understanding of patient decision-making around genome sequencing. *Personal Med* 15(1): 57–66
- Hylind R, Smith M, Rasmussen-Torvik L, Aufox S (2018) Great expectations: patient perspectives and anticipated utility of non-diagnostic genomic-sequencing results. *J Community Genet* 9(1):19–26
- Jacob H, Abrams K, Bick D, Brodie K, Dimmock D, Farrell M, Geurts J et al (2013) Genomics in clinical practice: lessons from the front lines. *Sci Translat Med* 5:194
- Kalia S, Adelman K, Bale S, Chung W, Eng C, Evans J, Herman G et al (2017) Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics. *Genet Med* 19(2):249–255
- Korol S, Hurlimann T, Godard B, deDenus S (2013) Disclosure of individual pharmacogenomics results in research projects: when and what kind of information to return to research participants. *Pharmacogenomics* 14:675–688
- Lemke A, Wu J, Waudby C, Somkin C, Trinidad S (2010) Community engagement in biobanking: experiences from the eMERGE network. *Genomics Society and Policy* 6:35.52
- Mackley M, Fletcher B, Parker M, Watkins H, Ormondroyd E (2017) Stakeholder views on secondary findings in whole genome and whole exome sequencing: a systematic review of quantitative and qualitative studies. *Genet Med* 19(3):283–293
- Manolio T, Chisholm R, Ozenberger B, Roden D, Williams M, Wilson R, Dick D et al (2013) Implementing genomic medicine in the clinic: the future is here. *Genet Med* 15:25–67
- Nunn J, Tiller J, Fransquet P, Lacaze P (2019) Public involvement in global genomics research: a scoping review. *Front Public Health* 7:79
- Pulley J, Denny J, Peterson J, Bernard J, Vnencak-Jones C, Ramirez A, Delaney J et al (2012) Operational implementation of prospective genotyping for personalized medicine: the design of the Vanderbilt PREDICT project. *Clin Pharmacol Ther* 92:87–95
- Roberts J, Robinson J, Diamond P, Bharadwaj A, Christensen K, Lee K, Green R, McGuire A, the MedSeq Project team (2018) Patient understanding of, satisfaction with, and perceived utility of whole genome sequencing: findings from the MedSeq Project. *Genet Med* 20(9):1069–1076
- Rosenman M, Decker B, Levy K, Holmes A, Pratt V, Eadon M (2017) Lessons learned when introducing pharmacogenomic panel testing into clinical practice. *Value in Health* 20:54–59
- Sandelowski M (2000) Whatever happened to qualitative description? *Res Nurs Health* 23(4):334–340
- Sandelowski M (2010) What's in a name? Qualitative description revisited. *Res Nurs Health* 33(1):77–84
- Schwartz M, McCormick C, Lazzeri A, Lindbuchler D, Hallquist M, Manickam K, Buchanan A et al (2018) A model for genome-first care: returning secondary genomic findings to participants and their providers in a large research cohort. *Am J Hum Genet* 103:328–337
- Shickh S, Clausen M, Mighton C, Casalino S, Joshi E, Glogowski E, Schrader K et al (2018) Evaluation of a decision aid for incidental genomic results, the Genomics ADvISER: protocol for a mixed methods randomized controlled trial. *BMJ Open* 8:e021876
- Vassey J, Christensen K, Schonman E, Blout C, Robinson C, Krier J, Diamond P et al (2017) The impact of whole genome sequencing on the primary care and outcomes of healthy adult patients: a pilot randomized trial. *Ann Intern Med* 167:159–169
- Wynn J (2016) Genomic testing: a genetic counselor's personal reflection on three years of consenting and testing. *J Genet Counsel* 25:691–697
- Yang Y, Muzny D, Reid J, Bainbridge M, Willis A, Ward P, Braxton A et al (2013) Clinical whole exome sequencing for the diagnosis of Mendelian disorders. *N Engl J Med* 369:1502–1511

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